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Noninvasive prenatal testing (NIPT)

What is NIPT?

NIPT, also known as cell-free DNA (cfDNA) screening, is a prenatal aneuploidy screening option. NIPT safely and noninvasively screens for the most common chromosomal aneuploidies as early as 10 weeks gestation, using a single maternal blood draw. Professional societies, including the American Congress of Obstetricians and Gynecologists (ACOG), have recommended NIPT as an option for common aneuploidy screening for all pregnant women, regardless of age or risk.[1,2]

NIPT can screen for:

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)
- Certain sex chromosome aneuploidies
- Select chromosomal microdeletions
- All autosomal trisomies
- Small extra or missing chromosomal changes on any chromosome

Screening options vary by laboratory. Talk to your laboratory provider about what is available for your patients.

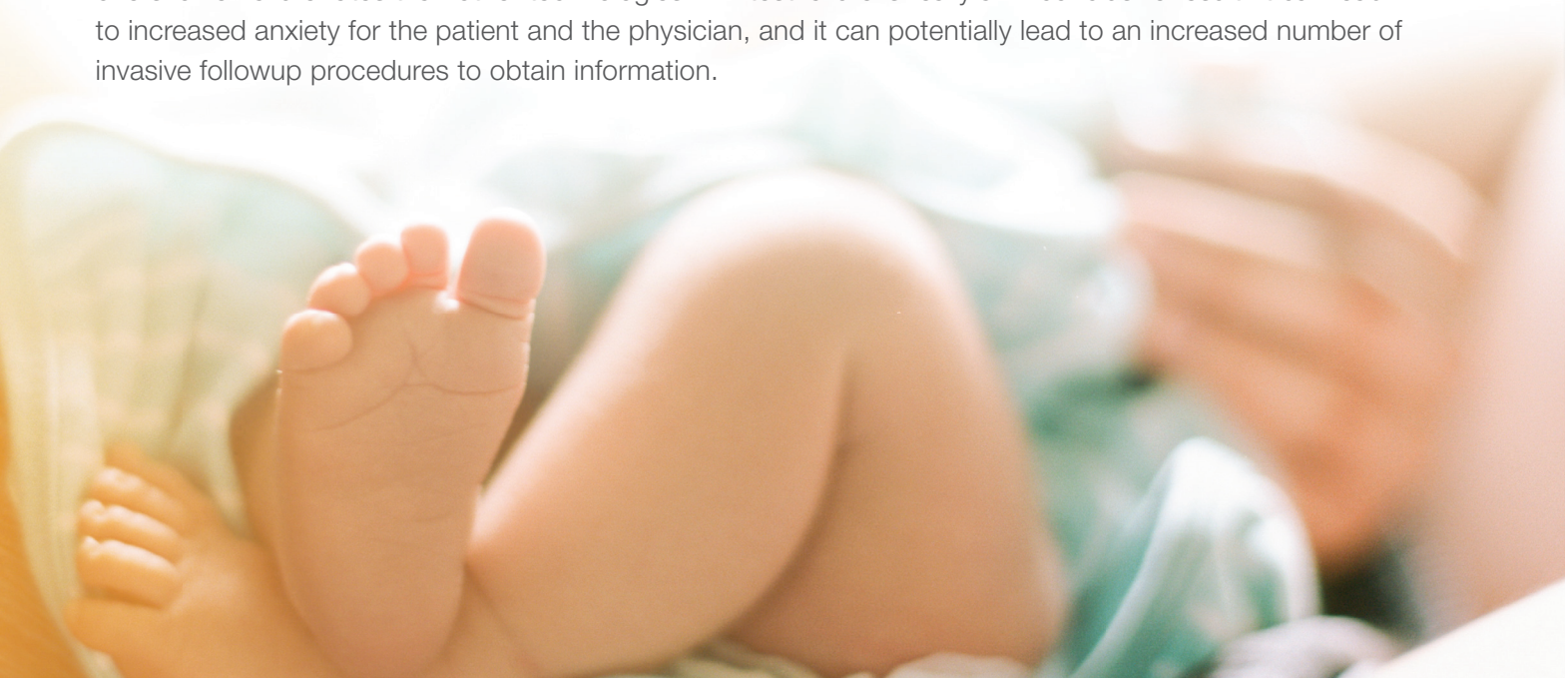
How does NIPT differ from traditional serum screening methods?

NIPT has a higher level of sensitivity and specificity than traditional serum screening[1–3] offering the:

- Highest reported detection rate for Down syndrome[1]
- Lowest reported false positive rate for Down syndrome[1]
- Broadest screening window (performed as early as 10 weeks gestation until term)[1–3]
- Ability to screen for additional chromosomal conditions [1-3]

How do I choose the best NIPT for my patients?

Choosing the right NIPT is important to your patients. While different NIPT methods are available, next-generation sequencing (NGS) is the most published method.[4] Whole-genome sequencing-based NIPT offers lower failure rates than other technologies.[5–9] A test failure is really an inconclusive result. It can lead to increased anxiety for the patient and the physician, and it can potentially lead to an increased number of invasive followup procedures to obtain information.



Limitations of the test

Noninvasive prenatal testing (NIPT) based on cell-free DNA analysis from maternal blood is a screening test; it is not diagnostic. False positive and false negative results do occur. Test results must not be used as the sole basis for diagnosis. Further confirmatory testing is necessary prior to making any irreversible pregnancy decision. A negative result does not eliminate the possibility that the pregnancy has a chromosomal or subchromosomal abnormality. This test does not screen for birth defects such as open neural tube defects, or other conditions, such as autism. Some NIPT tests do not screen for polyploidy (eg triploidy) or single gene disorders. There is a small possibility that the test results might not reflect the chromosomal status of the fetus, but may instead reflect chromosomal changes in the placenta (ie, confined placental mosaicism [CPM]) or in the mother that may or may not have clinical significance.

References

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